Chapter 1.1

History Highlights

Although this book is about neurological investigations, the best pointer to the appropriate tests and the correct diagnosis is the clinical history. The historian is the person taking the history, and this may take longer than the time allowed by many health systems.

A crucial aspect of the elicited biography is the situation, setting, state, stimulus and provocation. Like other aspects of the clinical history such details may need to be elicited in the sense of being squeezed out from the memory of the informants. Sometimes the existence of triggers may be denied, but if one allows undirected talk then a significant pointer may emerge. Although prolonged interrogation is often required, sometimes the simplest association gives the diagnostic clue. If an infant deteriorates shortly after DTP vaccine, then this is likely to be the start of Dravet syndrome with a mutation in the \textit{SCN1A} gene or a mitochondrial disorder or occasionally a metabolic decompensation, as in glutaric aciduria type 1 (GA1).

In the case of paroxysmal disorders it has been said that the “diagnosis is as good as the history” and this is true provided that the history is taken meticulously from the witness or witnesses to the event and from the child if sufficiently old. “The objective is to elicit a sequence which, replayed in the mind’s eye, is as good as or better than video recording with full polygraphy” (Stephenson 1990). Nonetheless, home video, with accompanying audio, may add to this moving picture, and even more so if such recordings are made with a specific question in mind (Chapter 2.1).

In subsequent chapters we include examples of specific approaches to history taking. As a very simple example, if the problem is delayed motor development, the first questions will focus on the style of developmental trajectory: is there a crawling sequence as is usual, or a bottom shuffling one (Chapters 3.3, 3.4, 3.7)? When the consultation relates to recently recognized learning difficulties (Chapter 3.9) the question is more difficult
and one must dissect the skills history to ensure that there has been no loss of abilities (Chapter 3.22) or even a rare treatable disorder (Chapter 3.23).

In several neurological disorders the family history may be difficult to elicit or completely denied. Although this might seem particularly common in certain paroxysmal disorders and channelopathies, it is a widespread problem. It may be necessary to interview other members of the family, particularly the oldest living female relative (the ‘matriarch’), to elicit the true state of affairs, and in several situations such as genetic epilepsy with febrile seizures plus (GEFS+) (Chapters 3.12, 3.13) this is essential.

If a diagnostic problem has not been solved by the sequence history examination targeted investigation(s), then one returns to the history and begins this process again.

We sometimes regard the Guthrie test (neonatal blood spot analysis) as an extension of the clinical history: although it was introduced to detect inborn errors of metabolism, it is invaluable as a record of congenital infections, especially cytomegalovirus (Chapter 2.10).

Figure 1.1.1 A section of cassette EEG/ECG recorded during a severe non-epileptic convulsive syncope in a neonate with hyperekplexia due to a dominant negative mutation in the GlyT2 gene \textit{SLC6A5}. Note that the ‘spikes’ – which are giant repetitive compound muscle action potentials (CMAP) – whether very rapidly recurring at 30Hz (‘tonic’) or at around 8Hz (‘clonic’), are confined to scalp areas overlying muscle and so are absent at the vertex (channels 2 and 5, arrowed). Similar repetitive CMAP ‘spikes’ are seen on the ECG channel. The EEG is virtually isoelectric because of the profound syncope, and for the same reason the ECG shows severe bradycardia with absent P waves and junctional escape rhythm. This appearance has only been reported in (neonatal) hyperekplexia.
Clinical vignettes

1.1.1 ‘Sporadic’ case
A boy had frequent episodes of grunting with intense stiffening and deep cyanosis from the neonatal period. He received a diagnosis of hyperekplexia (Fig. 1.1.1) and was kept in hospital for the first four months of his life. His parents had always denied any family history but many years later his mother revealed that she herself had been hospitalized for four months from the neonatal period. Family honour had prevented disclosure of this information previously. It turned out that both he and his mother had a pathogenic dominant mutation in the gene encoding the glycine transporter GlyT2.

1.1.2 You have got to see it
A schoolboy presented with a history that he would suddenly stiffen, particularly when he moved suddenly. These episodes would last a minute or so and then he was alright. It almost seemed from the way he described it as a sort of cramp, but he wasn’t able to describe it very well and his mother was rather reluctant to imitate his abnormal movements. In any event the boy was entirely normal; no abnormal movements could be reproduced. However, as the boy got out of his chair to leave he suddenly twisted his neck, stiffened his left arm and turned his body around in a grotesque posture which lasted 20 or 30 seconds and then stopped. The doctor immediately recognized paroxysmal kinesigenic dystonia, and asked the young man how often he had this problem and he said he had it two or three times a day. The doctor asked if it was a problem for him and he said it wasn’t much of a problem but it was embarrassing sometimes. Then his mother interjected and said, “Of course we would like him treated because it must be quite painful for him.” The doctor then asked her why she thought it was painful, if he himself hadn’t said so. The conversation then went something like this:

Mother – “It is obvious that it must be painful to be in such a position.”
Doctor – “But he didn’t say so himself.”
Mother – “But I know it must be.”
Doctor – “How do you know that?”
Mother – “Because when I had it it was very painful.”

The doctor stopped in his tracks: “When you had it?” he said to the mother.

Mother – “Yes – I had it from the age of 10 to about 23. It stopped when I had my children.”
Doctor – “What did the doctor think it was?”
Mother – “I don’t think I saw a doctor.”
Doctor – “What did your parents think about it?”

1 This is a Stuart Green vignette, reproduced from Deonna and Stephenson (2008), with permission.
Mother – “Well, it didn’t happen very often and I used to try and hide it with other movements. I never actually told my parents so nobody knew about it.”

Doctor – “So you are telling me you had this abnormal movement for 10 to 15 years and now it has gone away but you never mentioned it to me until now?”

Mother – “Well, I never really thought about it until we were talking about the subject and I really saw what happened to my son.”

He was put on carbamazepine and did very well.

**Comment**

For a variety of reasons family members don’t always tell you the critical information which gives the diagnosis.

**Further reading**


